EMD gene

emerin

Normal Function

The *EMD* gene provides instructions for making a protein called emerin. Although this protein is produced in many tissues, it appears to be particularly important for the normal function of muscles used for movement (skeletal muscles) and heart (cardiac) muscle.

Within cells, emerin is a component of the nuclear envelope. The nuclear envelope is a structure that surrounds the nucleus, acting as a barrier between the nucleus and the surrounding fluid (cytoplasm) inside the cell. The nuclear envelope has several functions, including regulating the movement of molecules into and out of the nucleus.

Emerin interacts with several other proteins on the inner surface of the nuclear envelope. Together, these proteins may be involved in regulating the activity of certain genes, controlling the cell division cycle, and maintaining the structure and stability of the nucleus. Emerin and related proteins also play a role in assembling the nucleus during the process of cell division.

Health Conditions Related to Genetic Changes

Emery-Dreifuss muscular dystrophy

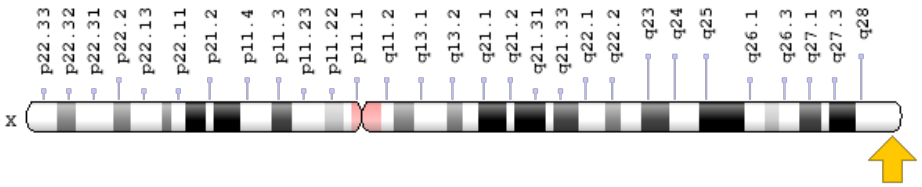
About 100 mutations in the *EMD* gene have been reported in people with Emery-Dreifuss muscular dystrophy. Almost all of these mutations prevent cells from producing any emerin protein. Researchers have not determined how a lack of this protein leads to the skeletal and cardiac muscle abnormalities characteristic of Emery-Dreifuss muscular dystrophy. Studies suggest, however, that an absence of emerin could disrupt the functions of other proteins in the nuclear envelope. These changes may alter the activity of certain genes or weaken the structure of the nucleus, making cells more fragile.

In rare cases, Emery-Dreifuss muscular dystrophy results from *EMD* mutations that change a single building block (amino acid) in the emerin protein. These mutations lead to the production of an abnormal version of emerin that is unable to interact with other proteins or cannot be correctly inserted into the nuclear envelope. This type of mutation may be responsible for some cases of Emery-Dreifuss muscular dystrophy with unusually mild signs and symptoms.

Chromosomal Location

Cytogenetic Location: Xq28, which is the long (q) arm of the X chromosome at position 28

Molecular Location: base pairs 154,379,237 to 154,381,523 on the X chromosome (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- EMD_HUMAN
- emerin (Emery-Dreifuss muscular dystrophy)
- STA

Additional Information & Resources

Educational Resources

- The Transport of Molecules between the Nucleus and the Cytosol (Molecular Biology of the Cell, Fourth Edition, 2002)
  https://www.ncbi.nlm.nih.gov/books/NBK26932/

GeneReviews

- Emery-Dreifuss Muscular Dystrophy
  https://www.ncbi.nlm.nih.gov/books/NBK1436

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28EMD%5BTIAB%5D%29+OR+%28emerin%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1440+days%22%5Bdp%5D

OMIM

- EMERIN
  http://omim.org/entry/300384

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_EMD.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=EMD%5Bgene%5D
- HGNC Gene Family: LEM domain containing
  http://www.genenames.org/cgi-bin/genefamilies/set/1087
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=3331
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/2010
- UniProt
  http://www.uniprot.org/uniprot/P50402

Sources for This Summary

- Bengtsson L, Wilson KL. Multiple and surprising new functions for emerin, a nuclear membrane protein. Curr Opin Cell Biol. 2004 Feb;16(1):73-9. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15037308
- Emery AE. Emery-Dreifuss muscular dystrophy - a 40 year retrospective. Neuromuscul Disord. 2000 Jun;10(4-5):228-32. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/10838246
- Fairley EA, Kendrick-Jones J, Ellis JA. The Emery-Dreifuss muscular dystrophy phenotype arises from aberrant targeting and binding of emerin at the inner nuclear membrane. J Cell Sci. 1999 Aug; 112 (Pt 15):2571-82. Erratum in: J Cell Sci 1999 Dec;112(Pt 24):following 4800.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/10393813
- GeneReview: Emery-Dreifuss Muscular Dystrophy
  https://www.ncbi.nlm.nih.gov/books/NBK1436
- Helbling-Leclerc A, Bonne G, Schwartz K. Emery-Dreifuss muscular dystrophy. Eur J Hum Genet. 2002 Mar;10(3):157-61. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/11973618

- Maraldi NM, Lattanzi G, Sabatelli P, Ognibene A, Squarzoni S. Functional domains of the nucleus: implications for Emery-Dreifuss muscular dystrophy. Neuromuscul Disord. 2002 Nov;12(9):815-23. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12398831
- Wilson KL, Holaska JM, Montes de Oca R, Tifft K, Zastrow M, Segura-Totten M, Mansharamani M, Bengtsson L. Nuclear membrane protein emerin: roles in gene regulation, actin dynamics and human disease. Novartis Found Symp. 2005;264:51-58; discussion 58-62, 227-30. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15773747

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